

When Baby Needs A Second Test for Classical Galactosemia Deficient GALT with Normal Total Galactose

A small sample of your baby's blood was collected soon after birth and sent to the DHEC laboratory for testing. This testing is called Newborn Screening. In SC, newborns are tested for several genetic and chemical disorders. Some times, a second test is needed to help your doctor decide if your baby has one of these disorders. In many cases, the second test will be normal. However, if your baby does have one of the newborn screening disorders, early treatment will give him or her the best chance to grow up healthy.

Because an enzyme called galactose-1-phosphate uridyl transferase (GALT) was low in your baby's first test, he or she could possibly have a disorder called classical galactosemia. Another compound, galactose, was also measured and was normal.

Please be aware that a lower than expected GALT can also be found in babies who do not have classical galactosemia. It is sometimes seen in babies with another form of galactosemia called Duarte galactosemia. Low GALT levels can also mean that your baby is a carrier of a galactosemia gene. Your baby needs to be retested quickly, though, as a precaution.

What is classical galactosemia?

Classical galactosemia is a genetic disorder that is found in a few babies born each year. It can be identified when an enzyme called GALT is measured in a baby's blood. When a baby has classical galactosemia, he or she cannot break down galactose, a part of the sugar lactose that is found in breast milk and cow's milk-based baby formula.

Galactose builds up in the baby's blood and damages the baby's body. The baby can have serious problems like swollen liver, dangerous infections, and brain damage.

What is Duarte galactosemia?

Duarte galactosemia is a mild form of galactosemia. Babies with Duarte galactosemia also have low amounts of GALT in their blood. However, there is enough GALT made by the baby so that the serious problems found in classical galactosemia do not happen.

How will I know if my baby really has galactosemia?

Since your baby's newborn screening result showed a low GALT level and a normal total galactose level, it is less likely that he or she has classical galactosemia. These kinds of results are more often seen when a baby has Duarte galactosemia or is a carrier for galactosemia. In any case, the newborn screening test will be repeated and additional tests may be done to help the doctors figure out if your baby has a form of galactosemia. Usually the results of these tests take a few days to come back. You may also be referred to a doctor who specializes in these kinds of disorders.

What do I need to do until I know the final results?

Since the total galactose was normal, you should not have to switch from breast milk or cow's milk-based formula to soy-based formula. Follow your doctor's instructions very carefully. If your baby is hard to wake up, refuses to eat, or seems to be getting sick, your doctor may have you call 911 for emergency help.

How is galactosemia treated?

Classical galactosemia is treated with a special diet. At first, babies with classical galactosemia must be fed a soy-based baby formula. When they begin to eat solids, the parents will have to be careful about which foods are given to the baby. The baby must not eat foods that contain any milk or dairy products, including all animals' milk like goat's milk. A dietitian will help the family learn which foods the baby can eat.

Babies with Duarte galactosemia are sometimes fed soy-based baby formula for the first year of life too. After that, babies with Duarte galactosemia can usually eat a regular diet.

What else should I do to keep my baby as healthy as possible?

Don't forget to keep all of your well baby check-ups! Seeing the doctors regularly and following your baby's diet plan carefully are the best things you can do to help your baby grow up healthy.